Recent Developments in Genetic Diagnosis: Some Ethical and Legal Implications

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The technology of genetic diagnosis, both in utero and after birth, is developing with breathtaking speed. It is already possible to diagnose prenatally such chromosomal abnormalities as trisomy 13, 18 and 21 (the last is known as Down's syndrome) and such genetic diseases as Tay-Sachs and sickle cell anemia. Within the very near future, reliable prenatal testing will likely be available for such genetic diseases as cystic fibrosis[1] and Huntington's chorea. In most cases, we will also be able to determine those individuals at risk of having offspring who suffer from these conditions, and further, those individuals who will develop later onset conditions, if they have not done so already. Genetic therapy is also appearing as a realistic possibility, particularly for diseases such as Lesch-Nyhan syndrome that are caused by a deficiency in a single gene.[2] These possibilities, developing every day, pose a number of ethical and legal problems for us both as individuals and as a society. If for no other reason than the speed of technological development, it is difficult for the law to keep pace.

This Essay outlines some of the ethical complexities genetic technology poses in two areas of decisionmaking: when to perform genetic testing and what to do with the information gained from genetic testing. My discussion does not promise any answers but an overview of why the issues are so complex.

A. The Decision to Perform Genetic Testing

As with the decision to perform other forms of diagnostic testing, the decision to perform genetic diagnosis involves a determination that medical intervention—albeit of a minimal diagnostic sort—is appropriate. Suppose we begin the search for criteria to

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1. *See, e.g.*, Newmark, *Testing for Cystic Fibrosis*, 318 NATURE 309 (1985).

2. *See* Areen, *Regulating Human Gene Therapy*, 88 W. VA. L. REV. 153, 159, n.32 (1985).

guide such decisionmaking with the standard that is common to other decisions about diagnosis and treatment: test when the benefits of testing outweigh the risks. This standard is complex enough in ordinary medical contexts. It involves at a minimum both physiological and psychological judgments.[3] In the context of genetic diagnosis this standard is particularly difficult, for a wide range of risks and benefits are involved for different people, including parents, offspring and society.

Consider first the case of prenatal testing through amniocentesis for a genetic disease such as Tay-Sachs or a chromosomal abnormality such as Down's syndrome. There are minor physical risks to both mother and fetus from the test procedure itself. Amniocentesis poses a slight risk of infection and a one in 200-300 risk of miscarriage. On the other side, however, there may be physical benefits for either mother or child in some cases in which prenatal diagnosis is possible. Treatment in utero of hydrocephalus, for example, may reduce neural damage.

Other benefits of amniocentesis go beyond the merely physical. The parents may benefit from knowing whether the fetus is afflicted by being relieved of worry or by being able to adjust to the coming birth of an impaired infant. They will also be able to decide on the basis of this prenatal test information whether to terminate the pregnancy. In addition, the availability of such information may clearly benefit the fetus. The fetus benefits if, for example, the mother would not have conceived the child, or would have terminated the pregnancy because of a genetic risk.

If a method of genetic diagnosis is less than fully accurate, or is inaccurately performed, however, there are risks for both parent and child that a pregnancy will be terminated in the mistaken belief that the fetus is affected. Most controversial of all are the possible consequences for the fetus of the mother's knowledge that the

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3. Courts in the United States have disagreed on the proper standard for informed consent by patients undergoing diagnostic procedures. 2 D. LOUISELL & H. WILLIAMS, MEDICAL MALPRACTICE ¶ 22.15-.65 (1985). The traditional standard is that the patient should be told about risks that the reasonable physician would disclose. *Id.* ¶ 22.06. This is still the rule in England and in many jurisdictions in the United States. Schwartz & Grubb, *Why Britain Can't Afford Informed Consent*, 15 HASTINGS CENTER REPORT, Aug. 1985. at 19, 21-22. Since the decision in Canterbury v. Spence, 464 F.2d 772 (D.C. Cir.), *cert. denied,* 409 U.S. 1064 (1972), however, an increasing number of jurisdictions in the United States have required disclosure of risks that the prudent patient would want to take into account in deciding whether to undergo a medical procedure. 2 D. LOUISELL & H. WILLIAMS, *supra*, ¶ 22.08. How far disclosure of these risks should be extended in the interest of patient autonomy, however, remains controversial. *See, e.g.*, Curran, *Informed Consent in Malpractice Cases*, 14 NEW ENG. J. MED. 429, 429-31 (1986).

fetus is affected by a genetic or chromosomal condition. Some argue that abortion is a benefit to a fetus affected by a condition such as Tay-Sachs, who faces a short period of development followed by potentially painful deterioration and death within two to five years. Others argue that it is not a benefit to a fetus afflicted with less severe or less rapidly fatal conditions such as Down's syndrome or Huntington's disease. Perhaps the most difficult of such choices involve conditions that permit the development of some years of meaningful life, coupled with suffering and early death. Cystic fibrosis is an example; many victims now survive until their late teens or early twenties, but must endure increasingly painful therapy and hospitalizations to preserve life. Still others argue, perhaps for religious reasons, that abortion is never a benefit to a fetus, no matter how grave the condition.

Now consider the risks and benefits to an individual of knowledge at any point before conception that he or she is at genetic risk of conceiving an afflicted child. One could argue on utilitarian grounds, or from the importance of preserving traditional familial institutions, that it is better to make decisions about marriage, procreation and other such basic life choices in blissful ignorance about genetic risks. If genetic diagnosis is not performed, some couples will encounter tragedies that they would prefer to have avoided, such as the birth of a doomed child. Others will know that they are at risk of tragedy because, for example, of other births or family events. They may as a result choose options (for example, remaining childless) they would not have chosen had genetic diagnosis been available. Still others may miss the possibility of therapeutic intervention, although this possibility is not available for currently untreatable genetic diseases such as Tay-Sachs. These risks are avoidable with genetic diagnosis.

Other risks, however, may present themselves when diagnosis is performed. For example, knowledge about their genetic risks may be a source of unhappiness to people who possess it. It may devastate others close to those at risk. Marriages may founder, or engagements be broken. Further, insurance companies and employers may use information about genetic risk to the disadvantage of those screened. These nonphysical risks from genetic information are serious indeed and will be discussed more fully in the section below.

The possibility of a mass screening program presents other nonphysical risks. Mass screening for sickle cell anemia, for example, was tentatively introduced but met with outcries that it could

have racist overtones if it encouraged abortions among blacks.[4]

The situation is complicated still further if we consider research aimed at developing new methods of genetic diagnosis. Physical risks of such research to subjects may be slight or nonexistent. In the usual case, diagnostic research involves the use of blood or tissue samples obtained for other purposes. There may be benefits from the research for the experimental subject, such as the diagnosis and treatment of active disease. Concomitantly, experimental diagnostic instruments risk inaccuracy; misinformation conveyed to subjects could be used in ways that are physically or psychologically damaging. For example, early efforts to develop a method of identifying carriers of cystic fibrosis raised high hopes of accuracy but proved disappointing.[5]

The risks and benefits of experimentation with genetic diagnosis also include, of course, the eventual development and use of the very tools of genetic diagnosis under experimental scrutiny. Moreover, experimentation may prove only a partial success. It may be possible, for example, to develop a test for the presence of active disease without being able to identify carriers or perform accurate prenatal diagnosis. Tests of carrier status may become available before tools for prenatal diagnosis. Individuals are then confronted with the knowledge that they are carriers, but that the condition of their particular unborn children cannot be ascertained.

B. Information Obtained in Genetic Diagnosis

The possibilities for use of information obtained in genetic diagnosis pose ethical dilemmas fully as troubling as the dilemmas posed by the decision to test. Genetic research generates these dilemmas most sharply, but they also appear in ordinary uses of genetic diagnosis.

First, conveying information to those being tested themselves poses both risks and benefits. The knowledge that at some point one will come to suffer from a genetic disease or the knowledge that one is at risk to have genetically impaired offspring are examples of the possible risks. These risks are magnified if the information is likely to be a surprise, as when an individual learns of some incidental condition other than that for which testing was initially

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4. PRESIDENT'S COMMISSION FOR THE STUDY OF ETHICAL PROBLEMS IN MEDICINE AND BIOMEDICAL AND BEHAVIORAL RESEARCH, SCREENING AND COUNSELING FOR GENETIC CONDITIONS 22-23 (1983) [hereinafter cited as PRESIDENT'S COMMISSION].

5. *Id.* at 89.

undertaken.

The risks of information are also magnified if the scientific validation of the diagnostic tool is still under study. Then there is some unknown likelihood that the information conveyed is inaccurate. Genetic counselors report that experimental subjects want to know the results of work in progress. The standard approach, however, is not to make results known until the scientific accuracy of a diagnostic procedure is established. This is the approach taken for genetic research protocols submitted to the University of Utah's Institutional Review Board, for example.[6]

Additional ethical problems arise if the genetic information has implications for other individuals, for example, an affected fetus or other members of a family tree. The potential risks and benefits for a fetus when parents are made aware of genetic diagnoses have been mentioned already. There are additional risks and benefits for family members who otherwise may not know that they have the potential for serious, genetically linked disease. In some cases, such as multiple polyposis of the colon (a precursor of colon cancer), availability of the information may make possible lifesaving early diagnosis. Yet tested individuals may have been assured confidentiality about test results[7] and may refuse to allow their relatives to be informed from fear of stigmatization or for other reasons. The President's Commission for the Study of Ethical Problems in Medicine recommended avoiding such situations when possible by insisting on disclosure of such potentially lifesaving information to relatives as a precondition of diagnostic testing itself. Recognizing that insistence on disclosure might discourage needed diagnosis or might simply not have been considered, however, the Commission also recommended guidelines for involuntary disclosure in other cases by analogy to disclosure of communicable disease. These guidelines were that reasonable efforts to obtain consent to disclosure had failed, that the probability is high that disclosure would avoid serious harm that would otherwise occur, and that precautions are taken to ensure disclosure only of the genetic information needed for diagnosis and treatment.[8]

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6. UNIVERSITY OF UTAH INSTITUTIONAL REVIEW BOARD, POLICIES AND PROCEDURES (1985).

7. In Utah, for example, medical records maintained by state institutions are classified as "private" data, available only to state agencies for appropriate uses, the individual himself, and others by the individual's express consent. UTAH CODE ANN. § 63-2-61(12) (Supp. 1985) and regulations.

8. PRESIDENT'S COMMISSION, *supra* note 4, at 44.

These guidelines were proposed for situations involving the use of nonexperimental techniques of genetic diagnosis. One of the hardest questions for genetics researchers, however, concerns whether information about experimentally tested subjects should be conveyed to other family members with important health interests. Researchers typically guarantee experimental subjects that identifying information about them will be kept confidential, and contacted subjects sometimes do not wish to have information about genetic risk conveyed to other family members. Indeed, the federal regulations governing research with human subjects require that experimenters inform subjects of their planned uses of data, and do not allow experimenters to make changes without the subjects' consent.[9]

These risks of information are perhaps most patent if we focus on the situation of an individual who is himself at risk of genetic disease. Identification of possessors of the Huntington's gene, for example, will relieve some and consign others to knowledge of their inevitable fates. The alternative would be to leave all members of a family in which Huntington's has appeared uncertain about their futures. Some may want to know; others will prefer the uncertainties of ignorance to the possibility of knowing their eventual dooms. But whether an individual chooses to find out about his susceptibility to genetic disease is not a decision that will affect him alone. Partners, offspring, even employers have much at stake in an individual's decision to acquire or decline relevant information about his or her genetic status.

Identification of susceptibility to genetic disorders may be used to deny insurance benefits or to limit employment opportunities. Screening for genetic susceptibility to toxins, for example, has recently been implemented by several major employers.[10] Concerns about the practice include questionable ways in which employers may use genetic information. For instance, such information may be used as a pretext for racial or sexual discrimination, or as a method to avoid remediating workplace conditions in favor of excluding hypersensitive workers. Genetic screening may provide employers with important information to improve workplace safety, however, and workers arguably have the right to be fully informed about potential hazards of their workplace environment, at least

9. 45 C.F.R. § 46.111(a)(7) (1985).
10. *See, e.g., Genes and Jobs*, 68 A.B.A. J. 1061, 1061 (1982).

some of which are surely individual.[11] Ensuring the privacy and confidentiality of workers screened in such programs poses additional legal difficulties.

C. *Legal Responses*

There are several possible legal responses to these ethical dilemmas. This discussion focuses on two. First, regulatory approaches have been proposed to the performance of genetic testing or the use of genetic information. Second, tort law has been used as a means to shift losses associated with the performance (or nonperformance) of genetic testing and the use of information gained in testing.

An example of the regulatory prohibition of genetic testing was the Food and Drug Administration's refusal to give market clearance until the summer of 1985 for blood test kits for diagnosing neural tube defects. The concern was that test information would be misused because of inadequate facilities for followup testing. The marketed kit reveals only the need for further testing to rule out a neural tube defect; it does not show the clear presence of a defect. Individuals with positive results but inadequate followup might be led thereby to terminate pregnancies precipitously.[12]

Regulatory approaches have also been proposed for the use of information obtained in genetic diagnosis. For example, in addition to its guidelines concerning involuntary disclosure of the results of genetic diagnosis to relatives, the President's Commission recommended that information about genetic diagnosis be conveyed to employers or insurers only with the patient's consent.[13] Very recently, the suggestion has been made that genetic screening with

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11. *See, e.g.*, Diamond, *Genetic Testing in Employment Situations*, 4 J. LEGAL MED. 231, 247-54 (1983); Goodrich, *Are Your Genes Right for Your Job?*, 3 CAL. LAW., May 1983, at 24, 27-28; Sweltz, *Genetic Testing in the Workplace: An Analysis of the Legal Implications*, 19 FORUM 323, 325-26 (1984); Note, *Getting Beyond Discrimination: A Regulatory Solution to the Problem of Fetal Hazards in the Workplace*, 95 YALE L.J. 577, 577-79 (1986).

12. In the summer of 1985, the American College of Obstetricians and Gynecologists reacted to the Food and Drug Administration's decision to grant market clearance for the screening kit. Their initial response was a legal recommendation that screening should now be part of the standard of care. The recommendation still stands, but the American College of Obstetricians and Gynecologists has attempted to "retreat without retracting." Annas, *Is a Genetic Screening Test Ready When the Lawyers Say It Is?*, HASTINGS CENTER REPORT, Dec. 1985, at 16.

13. PRESIDENT'S COMMISSION, *supra* note 4, at 42.

respect to toxic substances in the workplace be handled under the Toxic Substances Control Act rather than being regarded, as it largely has been, as a problem of sexual discrimination.[14]

A second legal approach with great potential for influencing the conduct of genetic diagnosis is loss shifting through tort liability. In the past few years, a number of states have recognized tort law suits on the theory of wrongful birth.[15] A wrongful birth suit is brought by the child's parents, who contend that but for the negligence of health care providers, their child would not have been born. Examples of such negligence include the failure to recognize the need for prenatal testing, the failure to inform parents of the availability of prenatal testing, or the careless performance of prenatal diagnosis. In the typical wrongful birth suit, the parents seek as damages the costs of raising the child whose birth would have been avoided. They may also seek recovery for their own pain and suffering resulting from the birth of the child. Although there is some division, courts are more likely to allow parents damages in situations involving the birth of an impaired child than in situations in which the birth of a healthy child follows practitioner negligence.[16] It is also more likely that parents will recover the costs of raising the child, which if the child is handicapped (as is common in these cases) may be quite high, than that the parents will re-

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14. *See* Note, *supra* note 11, at 591-96. The most important advantage of an approach through the TSCA is that it allows the problem of reproductive hazard to be approached comprehensively in a manner which considers risks to both men and women.

15. *See* Turpin v. Sortini, 31 Cal. 3d 220, 239, 643 P.2d 954, 966, 182 Cal. Rptr. 337, 339 (1982); Fassoulas v. Ramey, 450 So. 2d 822, 824 (Fla. 1984); Blake v. Cruz, 108 Idaho 253, 698 P.2d 315 (1984); Troppi v. Scarf, 31 Mich. App. 240, 187 N.W.2d 511, 513-14 (1971); Sherlock v. Stillwater Clinic, 260 N.W.2d 169, 170-71 (Minn. 1977); Berman v. Allen, 80 N.J. 421, 404 A.2d 8, 14 (1979); Becker v. Schwartz, 46 N.Y.2d 401, 386 N.E.2d 807, 813, 413 N.Y.S.2d 895 (1978); Bowman v. Davis, 48 Ohio St. 2d 41, 356 N.E.2d 496 (1976); Speck v. Finegold, 497 Pa. 77, 439 A.2d 110, 113-14 (1981); Jacobs v. Theimer, 519 S.W.2d 846, 850 (Tex. 1975); Harbeson v. Parke-Davis, 98 Wash. 2d 460, 656 P.2d 483, 488 (1983); Dumer v. St. Michael's Hosp., 69 Wis. 2d 766, 233 N.W.2d 372, 377 (1975). *But see* Azzolino v. Dingfelder, 315 N.C. 103, 337 S.E.2d 528, 533 (1985) (claims for wrongful birth should not be judicially recognized absent a clear legislative mandate).

16. *Compare* Smith v. Gore, No. CA-1006, slip op. at 2 (Tenn. App. Jan. 28, 1986) (costs of rearing a normal, healthy child are not recoverable, but nothing in opinion precludes recovery of proximate damages not included in cost of rearing the child), Garrison v. Foy, 486 N.E.2d 5, 10 (Ind. 1985) (possible damages limited to those directly caused by unsuccessful sterilization and do not include costs of raising child nor exceptional expenses associated with child's defect), *and* Beardsley v. Wierdsma, 650 P.2d 288 (1982) (refusing to allow parents the costs of raising a healthy child) *with* Ochs v. Borrellia, 445 A.2d 883 (Conn. 1982) (allowing parents the costs of raising a healthy child, offset by benefits from the child's existence).

cover for their own pain and suffering.[17]

Some states have also recognized a cause of action for what is called wrongful life. This is a suit brought on behalf of the child claiming that it would not have been born alive but for practitioner negligence.[18] The most common criticism of this cause of action is the difficulty of measuring damages. Courts rejecting wrongful life suits argue either that being born cannot be a wrong to a child, no matter what pain she suffers from her impairments, or that whatever these damages are they cannot be ascertained.[19] The extraordinary medical expenses associated with having a genetic defect are measurable, however, and at least one court has allowed a child to recover these damages, although not allowing recovery for the impaired existence itself.[20]

The Utah Legislature recently added a new chapter to wrongful birth and wrongful life litigation in the state. In 1983, the legislature passed the Utah Wrongful Life Act.[21] The Act's stated aim is "to encourage all persons to respect the right to life of all other persons, regardless of age, condition or dependency, including all handicapped persons and all unborn persons."[22] To this end, the Act specifies that no cause of action may arise in Utah based on the claim that but for the act or omission of another, a person would not have been born alive but would have been aborted.[23] It also stipulates that failure to prevent a live birth shall not be considered in awarding damages in any action.[24]

With one swoop, the Act abolishes both wrongful birth and

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17. *See* Becker v. Schwartz, 46 N.Y.2d 401, 386 N.E.2d 807, 813-14, 413 N.Y.S.2d 895 (1978). *But see* Berman v. Allen, 80 N.J. 421, 404 A.2d 8, 14 (1979) (allowing parents in a wrongful birth suit to recover). *See generally* Comment, *The Legal Recognition of Medical Malpractice Tort Claims Based Upon Theories of Wrongful Birth and Wrongful Life*, 15 N.C. CENTRAL L.J. 274, 280 (1985) (noting the difficulty of calculating damages for the parents' emotional pain and suffering as compared to presenting a verified list of medical expenditures).

18. *See, e.g.*, Turpin v. Sortini, 31 Cal. 3d 220, 237-39, 643 P.2d 954, 965-66, 182 Cal. Rptr. 337, 348-49 (1982); Curlender v. Bio Science Laboratories, 106 Cal. App. 3d 811, 830, 165 Cal. Rptr. 477, 479 (1980); Siemieniec v. Lutheran General Hosp., 134 Ill. App. 2d 823, 480 N.E.2d 1227, 1235 (1985); Procanik v. Cillo, 97 N.J. 339, 478 A.2d 755, 762 (1984); Harbeson v. Parke-Davis, Inc., 98 Wash. 2d 460, 656 P.2d 483, 493-95 (1983).

19. *See, e.g.*, Blake v. Cruz, 108 Idaho 253, 698 P.2d 315, 322 (1984).

20. Siemieniec v. Lutheran General Hosp., 134 Ill. App. 2d 823, 480 N.E.2d 1227, 1235 (1985).

21. Act of March 10, 1983, 1983 Utah Laws 687 (codified at UTAH CODE ANN. §§ 78-11-23 to -25 (Supp. 1985).

22. UTAH CODE ANN. § 78-11-23 (Supp. 1985).

23. *Id.* § 78-11-24.

24. *Id.* § 78-11-25.

wrongful life as causes of action in Utah. The Act probably also means that negligent prescription of contraceptives cannot be considered in awarding damages. The Act does not, to be sure, rule out all damages from negligence with respect to genetic testing. For example, individuals could recover if failure to test had resulted in the exacerbation of a genetic condition. But it does rule out one set of potentially large recoveries—those for the costs of raising a child who otherwise would not have been born—and it does this both in the case of couples who would have chosen to abort an affected fetus and in the case of couples who would have chosen not to conceive in the first place. The Act is thus far more than an anti-abortion statute, despite its announced right to life policy.[25]

Utah is nearly unique in prohibiting wrongful birth and wrongful life suits by statute. Only South Dakota appears to have gone as far or farther than Utah in prohibiting such causes of action. South Dakota prohibits actions or damage awards based on the claim that but for the conduct of another, a person would not have been conceived or would not have been permitted to have been born alive.[26] Minnesota ties the prohibition of wrongful birth and wrongful life suits explicitly to efforts to discourage abortion. The Minnesota statute prohibits wrongful life or birth suits based on the claim that, but for the negligent acts of another, a person would have been aborted; it goes on to specify explicitly that

> [n]othing in this section shall be construed to preclude a cause of action for intentional or negligent malpractice or any other action arising in tort based on the failure of a contraceptive method or sterilization procedure or on a claim that but for the negligent conduct of another, tests or treatment would have been provided or would have been provided properly which would have made possible the prevention, cure, or amelioration of any disease, defect, deficiency, or handicap; provided, however, that abortion shall not have been deemed to prevent, cure, or ameliorate any disease, defect, deficiency, or handicap.[27]

Illinois has announced by statute that it is state policy to protect the life of the unborn child from the moment of conception and that the permissibility in Illinois of abortions in cases in which the

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25. *See* Note, *Wrongful Birth and Wrongful Life: Analysis of the Causes of Action and the Impact of Utah's Statutory Breakwater*, 1984 UTAH L. REV. 833, 856-63.

26. S.D. CODIFIED LAWS ANN. § 21-55-1 (Supp. 1984).

27. MINN. STAT. ANN. § 145.424 (West Supp. 1986).

mother's life is not threatened is solely a function of decisions of the United States Supreme Court.[28] This statute, however, has not been interpreted to prohibit wrongful life or wrongful birth suits in Illinois, based on the theory that an impaired fetus would have been aborted but for negligent medical advice.[29] Finally, California has prohibited by statute suits by the child against its parents based on a theory of wrongful life. This does not prohibit actions, however, by parents or the child against negligent health care providers; indeed, California makes it clear that parental failure to prevent live birth shall not be considered as a defense or a limit on damage awards in any actions against third parties.[30]

Regardless of views about the morality of abortion, I hope I have said enough about the complex risks and benefits of genetic diagnosis to suggest that the broad sweep of the Utah Act is ill-advised. The Act has the potential to increase the risks of genetic diagnosis, far beyond achieving its primary goal of discouraging abortions.[31] In effect, the Act removes a major set of malpractice incentives to inform people appropriately about the availability of genetic diagnosis, to perform genetic diagnosis carefully and to communicate information about the results of genetic diagnosis accurately and understandably.

28. ILL. ANN. STAT. ch. 38, § 81-21 (Smith-Hurd Supp. 1985).

29. Siemieniec v. Lutheran General Hosp., 134 Ill. App. 3d 823, 480 N.E.2d 1227, 1232, 1234-35 (1985) (recognizing causes of action in Illinois for wrongful birth and wrongful life).

30. CAL. CIV. CODE § 43.6 (West 1982).

31. A simple but major step would be to amend the statute to include only section 1.